



CHEK2 gene

checkpoint kinase 2

Normal Function

The *CHEK2* gene provides instructions for making a protein called checkpoint kinase 2 (CHK2). This protein acts as a tumor suppressor, which means that it regulates cell division by keeping cells from growing and dividing too rapidly or in an uncontrolled way.

The CHK2 protein is activated when DNA becomes damaged or when DNA strands break. DNA can be damaged by agents such as toxic chemicals, radiation, or ultraviolet (UV) rays from sunlight, and breaks in DNA strands also occur naturally when chromosomes exchange genetic material.

In response to DNA damage, the CHK2 protein interacts with several other proteins, including tumor protein 53 (which is produced from the *TP53* gene). These proteins halt cell division and determine whether a cell will repair the damage or self-destruct in a controlled manner (undergo apoptosis). This process keeps cells with mutated or damaged DNA from dividing, which helps prevent the development of tumors.

Health Conditions Related to Genetic Changes

breast cancer

Li-Fraumeni syndrome

Although most cases of Li-Fraumeni syndrome are associated with mutations in the *TP53* gene, *CHEK2* gene mutations have been identified in several families with cancers characteristic of this condition. At least one family has a mutation that deletes a single DNA building block (nucleotide) at position 1100 in the *CHEK2* gene (written as 1100delC). The 1100delC mutation leads to the production of an abnormally short, nonfunctional version of the CHK2 protein. Researchers are uncertain whether *CHEK2* gene mutations actually cause Li-Fraumeni syndrome or are merely associated with an increased risk of several types of cancer, including those cancers often seen in Li-Fraumeni syndrome.

ovarian cancer

prostate cancer

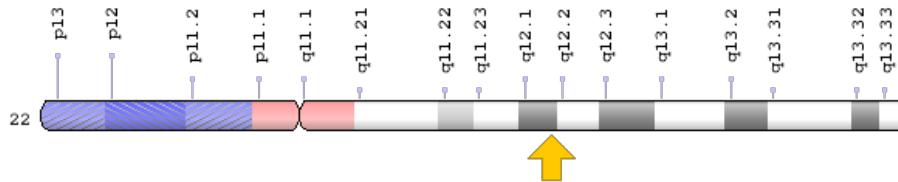
other cancers

Mutations in the *CHEK2* gene, including the 1100delC mutation described above, have also been found in other hereditary and nonhereditary (sporadic) cancers affecting many of the body's organs and tissues. Although the full range of cancers associated with *CHEK2* mutations has not been determined, studies have associated mutations in this gene with prostate, breast, lung, colon, kidney, thyroid, and ovarian cancers. *CHEK2* mutations have also been found in some brain tumors and in a type of bone cancer called osteosarcoma.

Chromosomal Location

Cytogenetic Location: 22q12.1, which is the long (q) arm of chromosome 22 at position 12.1

Molecular Location: base pairs 28,687,743 to 28,741,866 on chromosome 22 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CDS1
- Cds1 kinase
- checkpoint-like protein CHK2
- CHK2
- CHK2 checkpoint homolog (*S. pombe*)
- Chk2 protein kinase
- CHK2_HUMAN
- hCds1 protein
- hCHK2
- HuCds1

- RAD53
- serine/threonine-protein kinase CHK2

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Proto-Oncogenes and Tumor-Suppressor Genes
<https://www.ncbi.nlm.nih.gov/books/NBK21662/>
- National Cancer Institute: Genetics of Breast and Ovarian Cancer
<https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq>
- The Cell: A Molecular Approach (second edition, 2000): Tumor Suppressor Genes
<https://www.ncbi.nlm.nih.gov/books/NBK9894/>

GeneReviews

- Li-Fraumeni Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1311>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CHEK2%5BTIAB%5D%29+OR+%28CHK2+checkpoint+homolog%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- CHECKPOINT KINASE 2, S. POMBE, HOMOLOG OF
<http://omim.org/entry/604373>
- COLORECTAL CANCER
<http://omim.org/entry/114500>
- OSTEOGENIC SARCOMA
<http://omim.org/entry/259500>
- PROSTATE CANCER
<http://omim.org/entry/176807>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
<http://atlasgeneticsoncology.org/Genes/CHEK2ID312.html>
- **Cancer Genetics Web**
<http://www.cancerindex.org/geneweb/CHEK2.htm>
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=CHEK2%5Bgene%5D>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=16627
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/11200>
- **UniProt**
<http://www.uniprot.org/uniprot/O96017>

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<https://ghr.nlm.nih.gov/gene/CHEK2>

Reviewed: August 2007

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services